

COfiler STR(D3S1358, D16S539, TH01, TPOX, CSF1PO, D7S820)

4, 2, 3, 5, 1, 2, 5, 2, 4, 5, 3, 4, 5, 1, 2, 5, 1, 2, 5

= Abstract =

A Korean population study for the COfiler STR loci (D3S1358, D16S539, TH01, TPOX, CSF1PO, D7S820)

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In the United States, the Federal Bureau of Investigation (FBI) officially launched its national DNA database. This database, named the combined DNA Index System (CODIS), included one gender-determining amelogenin and 13 polymorphic short tandem repeats (STR) loci. To introduce a new STR system, a population database for the relevant population must be established for the statistical analysis of forensic cases. AmpFISTR Profiler Plus PCR Amplification Kit (Profiler Plus Kit) and AmpFISTR COfiler PCR Amplification Kit (COfiler Kit) are required to obtain information from all the 13 CODIS core STR loci. Study on 9 STR loci using Profiler Plus kit was already performed in a Korean population, but not yet on 6 STR loci using COfiler Kit.

This study intends to evaluate usefulness of 6 COfiler STR loci (D3S1358, D16S539, TH01, TPOX, CSF1PO, D7S820) in forensic identification. Buccal swab samples obtained from 300 randomly selected unrelated Koreans. DNA was extracted from the buccal swab samples and multiplex polymerase chain reaction (PCR) was performed using the COfiler Kit to amplify it. And using automated DNA sequencer and computer program, the allele and genotype frequency distribution is investigated and statistical analysis was performed for the PCR products. The following results were obtained:

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VNTR DNA RFLP - (laser - fluorescence detection)
 DNA , Southern blot, STR
 15, 16)
 7). RFLP 1998
 500 ng Combined DNA Index System(CODIS)
 DNA가 , , DNA FBI
 DNA가 amelogenin 13 STR
 D3S1358, VWA, FGA, D8S1179, D21S11,
 D18S51, D5S818, D13S317, D7S820, D16S539,
 TH01, THOX, CSF1PO
 Mullis Faloona⁹⁾
 (Polymerase chain reaction; PCR)
 Applied Biosystems AmpFISTR Profiler
 Plus PCR Amplification Kit (Profiler Plus Kit)
 AmpFISTR COfiler PCR Amplification Kit(
 COfiler Kit) 가 .
 VNTR 가 ampli -
 fied fragment length polymorphism (AMP - FLP)
 DNA 가 가
 Craig ¹⁰⁾
 가 2 - 6 short tandem repeats
 (STR) , Litt Luty¹¹⁾
 microsatellite . STR
 VNTR STR
 가 가
 Profiler Plus Kit 9
 (multiplex PCR) ¹⁷⁾, COfiler Kit
 VNTR STR
 (0.5 - 2 ng) ,
 가 STR
 Smith ¹³⁾ 가 ¹²⁾
 (primer) STR
 lane PCR FBI
 , Carrano ¹⁴⁾ 6
 DNA (internal size standard)
 lane
 DNA
 DNA 1.
 , 1991 STR (fluorescent STR
 markers)가 ,
 STR 가 300

, DNA
 2 - 3
 1.5 ml microcentrifuge tube
 DNA - 20
 (internal size standard) GeneScan - 500
 ROX(Applied Biosystems, Foster City, USA) 1.0
 $\mu\ell$ 24 $\mu\ell$ formamide
 95 3
 3
 2. (AmpFISTR COfiler Allelic Ladder; COfiler
 Allelic Ladder; Applied Biosystems, Foster City,
 USA) 1.0 $\mu\ell$
 3. DNA
 QIAamp DNA Mini Kit (QIA -
 GEN, Hilden, Germany)
 DNA 400 $\mu\ell$ phosphate
 buffered saline(PBS), 20 $\mu\ell$ QIAGEN protease,
 400 $\mu\ell$ Buffer AL 1.5ml
 microcentrifuge tube 15
 vortexing . 56 10
 400 $\mu\ell$
 100% 가 vortexing
 700 $\mu\ell$
 QIAamp spin column 6000×
 g 1
 . 500 $\mu\ell$ Buffer AW1 6000×g
 1 , 500 $\mu\ell$ Buffer AW2
 20000×g 3
 20000×g 1
 150 $\mu\ell$ Buffer AE 1
 6000×g 1
 - 20
 (polymerase chain reaction;
 PCR) 2 - 10 ng DNA AmpFISTR
 COfiler PCR Amplification Kit(Applied Biosystems,
 Foster City, U.S.A.)
 1/4 13 $\mu\ell$
 GeneAmp 9600
 PCR system(Perkin - Elmer, U.S.A.) 95
 11 94 1 , 59
 1 , 72 1 30
 60 45 4
 (allele frequency)
 (genotype)
 PowerStat Excel Template²³⁾
 24)
 (observed het -
 erozygosity), (expected heterozy -

gosity), (polymorphism information content; PIC), (power of discrimination; PD), (mean exclusion chance; MEC)

가 Ronald ²⁸⁾

Hardy - Weinberg 가 Hardy -

1) (가) (observed heterozygosity) (het - erozygote) PowerStat Excel sufflings) Weinberg Lewis Genetic Data Analysis(GDA) Fisher's exact test(based on 100000 Template²³⁾

() (expected heterozygosity) 300 COfiler STR (allele frequency) Nei Roychoudhury²⁵⁾ (genotype)

2) (polymorphism information content; PIC) 가 index 1. 가. D3S1358 D3S1358 12, 13, 14, 15, 16, 17, 18, 19 8 15 가 0.405 가 16 (0.292), 17 12, 13, 19 0.003 가 D3S1358 가 3, 가 14 17 (Fig. 1, Table 1).

3) (power of discrimination; PD) (0.210) 26) PowerStat Excel Template²³⁾ 27) D16S539 D16S539 8, 9, 10, 11, 12, 13, 14 7 9 (0.257) 가 가 11 (0.255), 12 (0.225), 8 (0.002) 가 D16S539 (mean exclusion chance; MEC)

가 5 , 가 13 . TH01
 18 가 4 , 가 12
 (Fig. 1, Table 2). 16 (Fig. 2,
 Table 3).
 . TH01
 TH01
 , 6, 7, 8, 9, 9.3, 10
 6 , 9
 가 0.518 가
 , 7 (0.218), 6 (0.167)
 , 10 0.018 가
 . TPOX
 TPOX
 , 8, 9, 10, 11, 12, 13
 6 , 8
 가 0.500 가
 , 11 (0.305), 9 (0.117)

Table 1. Allele frequency distribution of the D3S1358 locus in a Korean population

Allele	12	13	14	15	16	17	18	19
12	0							
13	0	0						
14	0	1	0					
15	0	0	10	49				
16	2	1	8	63	25			
17	0	0	1	54	42	12		
18	0	0	0	17	8	5	0	
19	0	0	0	1	1	0	0	0
Observed No.	2	2	20	243	175	126	30	2
Frequency	0.003	0.003	0.033	0.405	0.292	0.210	0.050	0.003

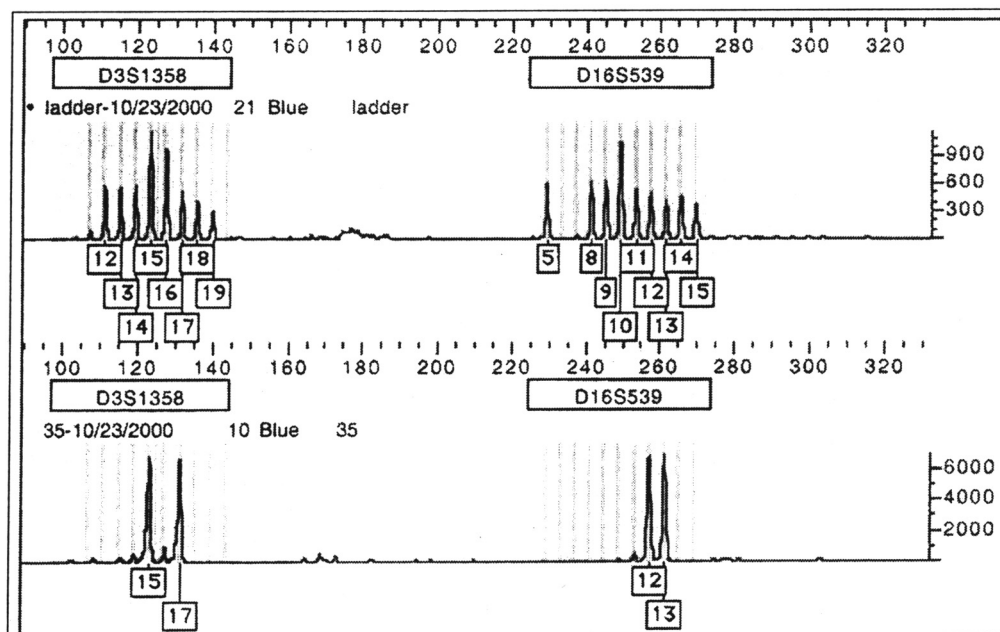


Fig. 1. AmpFISTR COfiler multiplex STR data for D3S1358 and D16S539 loci.

, 13 0.003 가 , 10 (0.250), 11
. TPOX (0.225) , 8 0.002
가 3 , 가 11 가
. CSF1PO
가 5 ,
14 (Fig.
2, Table 4). 가 15 20
(Fig. 2, Table 5).
. CSF1PO
CSF1PO . D7S820
7, 8, 9, 10, 11, 12, 13, D7S820
14 8 , 7, 8, 9, 10, 11, 12, 13,
12 가 0.403 가 14 8 .

Table 2. Allele frequency distribution of the D16S539 locus in a Korean population

Allele	8	9	10	11	12	13	14
8	0						
9	0	20					
10	0	30	1				
11	0	38	20	22			
12	0	34	23	31	13		
13	1	9	10	18	21	4	
14	0	3	0	2	0	0	0
Observed No.	1	154	85	153	135	67	5
Frequency	0.002	0.257	0.142	0.255	0.225	0.112	0.008

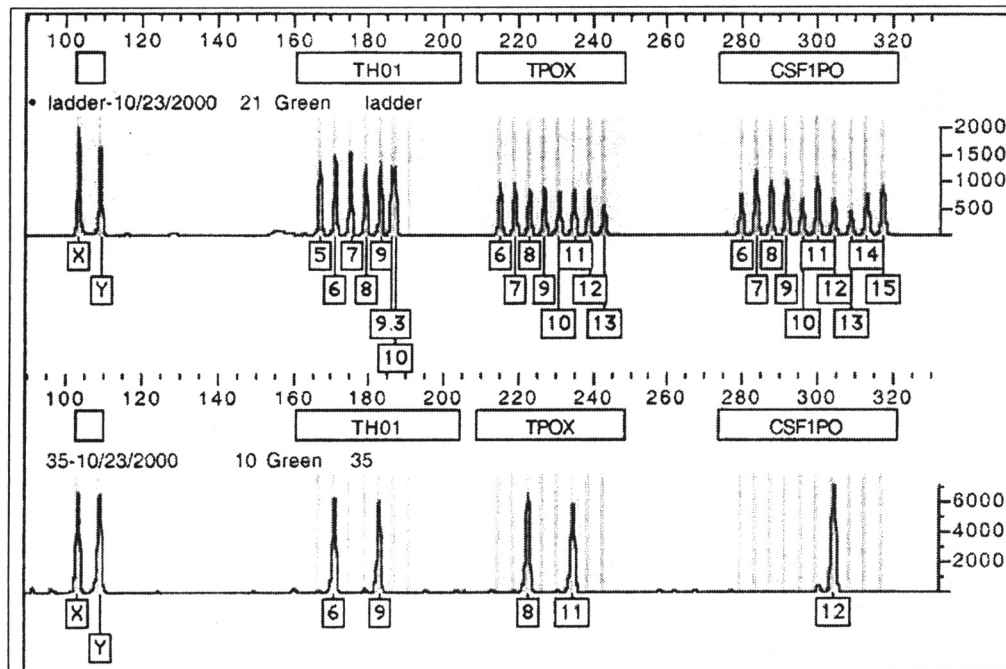


Fig. 2. AmpFISTR COfiler multiplex STR data for TH01, TPOX and CSF1PO loci.

11 가 0.338 가 D3S1358, D16S539, TH01, TPOX, CSF1PO, D7S820 (0.188), 8 (0.153), 12 (0.223), 10 (Table 7).

7 0.002 가 . (Fig. 3, Table 6).

가 5 , 가 19 24 3. Hardy - Weinberg Lewis GDA program Fisher's exact test(p) (based on 100000 suf- flings) D3S1358, D16S539, TH01, 2.

Table 3. Allele frequency distribution of the TH01 locus in a Korean population

Allele	6	7	8	9	9.3	10
6	8					
7	25	15				
8	3	3	0			
9	52	66	12	80		
9.3	4	5	0	18	0	
10	0	2	0	3	2	2
Observed No.	100	131	18	311	29	11
Frequency	0.167	0.218	0.030	0.518	0.048	0.018

Table 4. Allele frequency distribution of the TPOX locus in a Korean population

Allele	8	9	10	11	12	13
8	75					
9	28	3				
10	10	5	0			
11	95	26	4	26		
12	16	5	0	5	0	
13	1	0	0	1	0	0
Observed No.	300	70	19	183	26	2
Frequency	0.500	0.117	0.032	0.305	0.043	0.003

Table 5. Allele frequency distribution of the CSF1PO locus in a Korean population

Allele	7	8	9	10	11	12	13	14
7	0							
8	0	0						
9	0	0	2					
10	1	1	6	14				
11	0	0	2	31	12			
12	1	0	7	67	65	43		
13	0	0	2	13	10	16	1	
14	0	0	0	3	3	0	0	0
Observed No.	2	1	21	150	135	242	43	6
Frequency	0.003	0.002	0.035	0.250	0.225	0.403	0.072	0.010

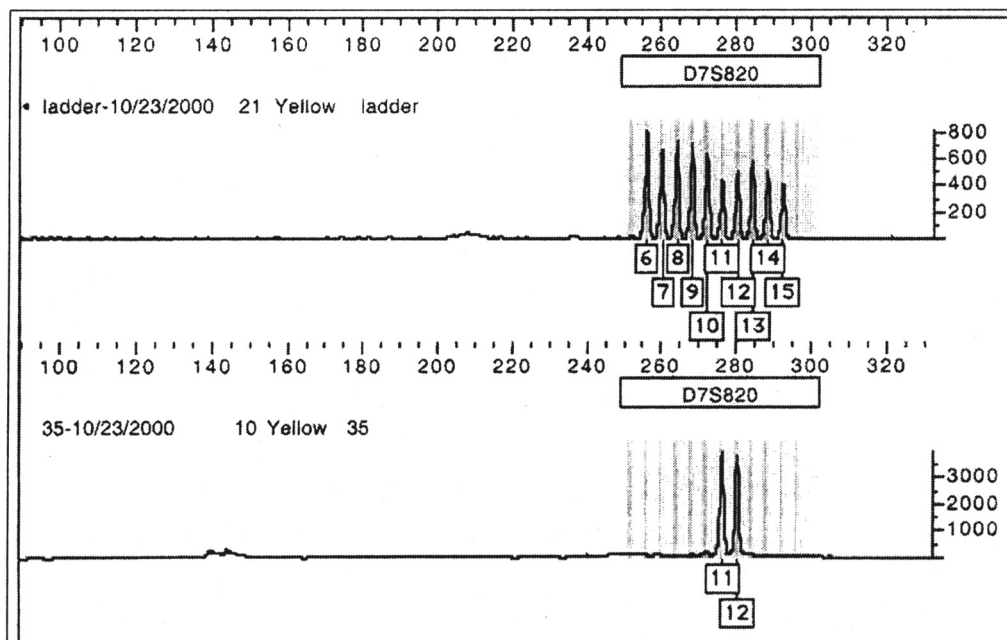


Fig. 3. AmpFlSTR COfiler multiplex STR data for D7S820 locus.

Table 6. Allele frequency distribution of the D7S820 locus in a Korean population

Allele	7	8	9	10	11	12	13	14
7	0							
8	0	4						
9	0	5	1					
10	0	19	6	8				
11	1	30	12	46	33			
12	0	25	6	21	40	18		
13	0	4	1	5	7	5	0	
14	0	1	0	0	1	1	0	0
Observed No.	1	92	32	113	203	134	22	3
Frequency	0.002	0.153	0.053	0.188	0.338	0.223	0.037	0.005

Table 7. Statistical parameters for the COfiler STR loci

Locus	D3S1358	D16S539	TH01	TPOX	CSF1PO	D7S820
obs-H ^a	0.713	0.800	0.650	0.653	0.760	0.787
exp-H ^b	0.704	0.787	0.653	0.642	0.719	0.774
PIC ^c	0.651	0.752	0.607	0.583	0.671	0.738
PD ^d	0.865	0.921	0.837	0.811	0.864	0.915
MEC ^e	0.452	0.576	0.415	0.386	0.477	0.562

^aobs-H: Observed heterozygosity, ^bexp-H: Expected heterozygosity

^cPIC: Polymorphic information contents, ^dPD: Power of discrimination, ^eMEC: Mean exclusion chance

TPOX, CSF1PO, D7S820 (automated DNA sequencer)

Hardy - Weinberg

가 lane

가 1ng DNA (inter -

가 .

nal size standard)

, 1bp

2bp DNA

가 400 - 1000bp VNTR

가

RFLP STR

amplified fragment length polymor -

phism(AMP - FLP)

(500 ng - 10 μ g) DNA 가 ,

(hybridization)

가 100 - 400

bp 가 , 가 DNA

(10ng)

DNA 가 가 STR

가

가 가

VNTR STR , STR

(Allelic Ladder)

AMP - FLP

DNA coding region noncoding region

가

coding region human genomic

DNA markers) DNA (hypervariable

DNA 5% noncoding region

VNTR STR ,

(multilocus probe) RFLP non - coding region

30 - 40%

human DNA

STR 가 , STR genome

10kb

30)

가 , PCR STR

5 - 6

(multiplex PCR)

(simple repeat locus),

(primer) 가 (com -

orescent - based typing) (flu - pound repeat locus),

(intervening

sequence) (complex (average random match probability) 1
repeat locus) ¹²⁾ 1 ³²⁾
STR 가 STR multiplex kit
(complete repeat units) 가 , PowerPlex 1.1 Kit PowerPlex
2.1 Kit (Promega Corporation GenePrint STR
Kits) Profiler Plus Kit COfiler Kit (Applied
Biosystems AmpFISTR Kits) 13
units) (incomplete repeat microvari -
ants TH01 STR
9.3 nucleotide 4 9
nucleotide 3 1
AATG 7
³¹⁾ adenine
amelogenin 6
가 가
, 가
Kit 가 Profiler
Plus Kit COfiler Kit D3S1358
D7S820 가
STR din -
nucleotide, trinucleotide, tetranucleotide, pentanu -
cleotide, hexanucleotide repeats
tetranucleotide repeats가 가
strand slippage
가 1 bp
, shutter
results ³³⁾ STR tetranucleotide
repeats (allele product
quantity) 15% , din -
nucleotides, trinucleotides 30%
. tetranucleotides dinucleotides,
trinucleotides shutter product
, (size - based
electrophoretic separations) 가
(heterozygotes)
가
³⁴⁾ 가
tetranucleotide repeats가 가
13 CODIS tetranu -
cleotide repeats .
(al allele frequen -
cy), (genotype frequency),

(heterozygosity) .

D3S1358
(Brazilian)
11, 20, 21 가 11
36), (African American),
(Western Australian), (Canadian
Caucasian) 8
37 - 39), (Andalusia),
(U.S. Caucasian), (Maghreb),
(Swiss Caucasian), (Hispanic)
12 7 40 - 43),
(Native Alaska) 12, 13, 19
5 43).

STR
0.9 가
70%
(closely linked
loci)가
(robustness) (reproducibility)
shutter product
(mutation rate) , DNA
90 - 500 bp
(allele length) 가 35).

(population genetics)
COfiler kit D16S539 16 long arm
6
- GATA . COfiler Allelic Ladder 5, 8, 9, 10,
11, 12, 13, 14, 15 9
가 ,
8, 9, 10, 11, 12, 13, 14 7 .
가 28
D3S1358 3 short arm
AGAT AGAC
. COfiler Allelic Ladder 가 5 , 가 13 18
12, 13, 14, 15, 16, 17, 18, 19 8 21) D16S539
가 ,
8 가 .
가 36 가
가 3 , 가 14 17
9 ,

, , , 5 가 7 .

5 8 , , , , , , , 6

, 5, 15 7 , 6

5, 8, 15 6 , 10 5

D16S539 TH01

9 , 11 가 가 9 가 가

21) , 20) , 19)

11 가 12 가 가 9 가 가

, , , 6 가

, , , , , , , , ,

11 가 , 7 가 , 9.3 가

12 가 , 9.3 가

Athabaskan 9가

10 , Inupiat, Yupik 11 가

, 11 12

, 9

TPOX 2 short arm

human thyroid peroxidase gene intron 10

TH01 11 short arm

tyrosine hydroxylase gene intron 1

TC11

HUMTH01

TCTA . COfiler Allelic Ladder 5, 6, 8,

7, 8, 9, 9.3, 10 7 가 , 8, 9, 10, 11, 12, 13 6 . 가

가 , 6, 21

7, 8, 9, 9.3, 10 6 .

가 21 가 3 , 가 11 14

가 4 , 가 12 16 20 13 5

19)

6 가 가 3 , 가 9 12

가 , 가 10 14

20)

9.3 5 가 , 가 9 6, 7 가 8 ,

13 . 13 6, 7

TH01 7

11 가 13 7 6

7 44, 45) , 8.3 , 13

가 7 , 14가 6 , 13

, , , 13 6 6

. , CSF1PO
 13 12 가 가
 5 20
 TPOX , , ,
 8 가 가 , , 가 12
 20) , 가 가 11
 , , , , 12 가 ,
 , 가 11 가
 8 가 가 .
 9 가 8 11 가 , 13, 12, 10
 11 가 8 D7S820 7 long arm
 CSF1PO 5 long arm GATA microvariant
 CSF - 1 receptor c - fms proto - onco - 가 . COfiler Allelic Ladder 6,
 gene 7, 8, 9, 10, 11, 12, 13, 14, 15 10
 AGAT . COfiler Allelic Ladder 6,
 7, 8, 9, 10, 11, 12, 13, 14, 15 10 가 , 7, 8, 9, 10, 11, 12, 13, 14 8
 가 , 가
 7, 8, 9, 10, 11, 12, 13, 14 8 36 ,
 가 가 5 ,
 36 , 가 19 24 D7S820
 가 15 20 가 5 ,
 20) 6 가 9 , , ,
 7 7 가 , , , 8
 가 5 , 가 13 7
 18 .
 CSF1PO 7 , ,
 10.3, 15 14 7 .
 가 가 10 D7S820
 15가 가 12 가 가 8 가
 6 가 9 , , , ,
 7 10.3, 가 , , , ,
 15가 9 , , , , ,
 , 10 가 ,
 8 , 8 10 가
 15가 가 8 11 .
 , 7 Athabaskan, Inupiat, Yupik
 15가 8 . 10 , 11 , 8 가
 7 7 .
 20

가 2
가

가 STR VNTR STR

가 , STR 2 3

가 3

가 Botstein²⁶⁾ 0.5

가 6 0.5

가 3 5

가 TPOX 가 6

가 , D16S539 가 가

가 6 D16S539 가 가

가 STR

가 A, B, AB, O 4가

가 ABO

가 ABO

가 STR

가 0.811 0.921

가 (1999)¹⁷⁾ Profiler Plus Kit

가 0.876

가 0.966

가 0.999996 COfiler STR

가 가

가 가

가 Budowle³⁶⁾ 6

가 (African American) 0.886 0.923

가 0.99999994

가 (U.S. Caucasian) 0.793 0.933

가 0.999998

가 6 STR

가 STR

가 2

가 (paternity test)

가 2

가 가

가 , ‘ ,

가 가 가 1. 0.650

가 0.800 , 0.642 0.787

가 가 2. 0.583

가 0.752 , 0.5

0.576 STR 0.386 3. 0.811

0.921 , 0.999996

0.98088 , Profiler Plus Kit 4. 0.386

17) 0.576 ,

(0.99983) 0.98088

Budowle³⁶⁾ (African , COfiler kit

American) COfiler STR 6 STR

0.511 0.603

0.99292

(U.S. Caucasian)

0.377 0.616

0.99055

6 STR

6

CODIS 13 STR

COfiler STR kit 6

STR (D3S1358, D16S539, TH01, TPOX, CSF1PO, D7S820)

가 300

DNA , COfiler

STR kit

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